

ABSTRACT

Research on family-based detection as a public health strategy for early detection of hemochromatosis.

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The disease hemochromatosis, a disorder of iron metabolism, occurs as a result of excess iron accumulation in tissues and organs. If left undiagnosed and untreated, iron overloading can cause serious health problems, and can even be fatal. In the US, it is estimated that as many as one in every 200 to 500, or approximately one million people, have evidence of hemochromatosis, and as many as one in every ten people may carry the genetic mutation for this disease. Hemochromatosis can be detected with simple blood tests and the treatment, phlebotomy, is relatively easy and inexpensive. Early diagnosis and treatment of hemochromatosis provides a tremendous opportunity to reverse the course of the illness and to prevent the most serious health problems of advanced stage hemochromatosis. At this time the most practical strategies for early diagnosis and treatment of hemochromatosis are enhanced case detection among individuals with hemochromatosis symptoms and family-based detection. Family-based detection is accomplished through (1) hemochromatosis patients urging their genetically-related family members to have their iron tested; and (2) physicians testing the iron of patients who have an increased risk for hemochromatosis because a family member has the disease, ascertained by family history. Therefore, it is important to understand factors that may promote or reduce the effectiveness of this family history-based approach. The CDC is currently conducting a research study among patients, siblings, and clinicians to understand family-based detection as a strategy to identify people with hemochromatosis.